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(54) Title: RESTRICTED AMPLICON ANALYSIS

(57) Abstract

The present invention generally provides a method which facilitates the detection of polymorphisms (or mutations). The method is directed to the analysis of so-called endonuclease site polymorphisms (ESPs) that result in the gain or loss of a restriction endonuclease site. In essence, the ESP is probed with the restriction endonuclease reagent prior to amplification, whereby amplification is prevented and consequently no signal is observed when cleavage takes place. Unambiguous allele calling is performed by comparing the signals obtained with and without cleavage with the restriction endonuclease reagent. The method is particularly useful for multiplex genotyping, involving the parallel analysis of large numbers of single nucleotide polymorphisms. Preferred methods for detecting the amplicons involve hybridization to an arrayed or otherwise identifiable set of cognate probe fragments or oligonucleotides.